



## SETX gene

senataxin

### Normal Function

The *SETX* gene provides instructions for making a protein called senataxin. Senataxin is produced in a wide range of tissues, including the brain, spinal cord, and muscles. Based on the structure of senataxin, researchers believe that it is one of a class of proteins called helicases that are involved in DNA repair and the production of RNA, a chemical cousin of DNA. Helicases attach to particular regions of DNA and temporarily unwind the two spiral strands of a DNA molecule. By unwinding the strands near sites of DNA damage, helicases allow other proteins to reach damaged areas and fix them. Senataxin is thought to aid in DNA repair through this helicase mechanism. This unwinding mechanism may also be involved in using the instructions encoded by RNA to create different versions of certain proteins (RNA processing).

### Health Conditions Related to Genetic Changes

amyotrophic lateral sclerosis

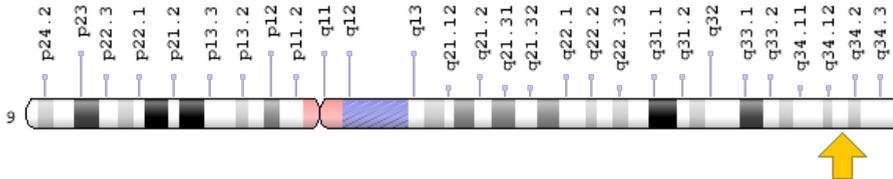
ataxia with oculomotor apraxia

At least 115 mutations in the *SETX* gene have been found to cause ataxia with oculomotor apraxia type 2. This condition is characterized by difficulty coordinating movements (ataxia) and problems with side-to-side movements of the eyes (oculomotor apraxia). Most mutations replace single amino acids in senataxin. The mutations associated with ataxia with oculomotor apraxia type 2 are thought to disrupt the helicase function of senataxin. A lack of functional senataxin disrupts DNA repair and can lead to an accumulation of DNA damage in cells, particularly affecting brain cells in the part of the brain involved in coordinating movements (the cerebellum). This accumulation can lead to cell death in the cerebellum, causing the characteristic movement problems of ataxia with oculomotor apraxia type 2.

## Chromosomal Location

Cytogenetic Location: 9q34.13, which is the long (q) arm of chromosome 9 at position 34.13

Molecular Location: base pairs 132,261,440 to 132,356,726 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- ALS4
- AOA2
- KIAA0625
- SCAR1
- SETX\_HUMAN

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Special Proteins Help to Open Up the DNA Double Helix in Front of the Replication Fork  
<https://www.ncbi.nlm.nih.gov/books/NBK26850/#A774>
- Molecular Biology of the Cell (fourth edition, 2002): The Structure of a DNA Helicase  
<https://www.ncbi.nlm.nih.gov/books/NBK26850/?rendertype=figure&id=A776>
- The Cell: A Molecular Approach (second edition, 2000): DNA Repair  
<https://www.ncbi.nlm.nih.gov/books/NBK9900/>
- Washington University, St. Louis Neuromuscular Disease Center  
<http://neuromuscular.wustl.edu/synmot.html#als4>

### GeneReviews

- Amyotrophic Lateral Sclerosis Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1450>
- Ataxia with Oculomotor Apraxia Type 2  
<https://www.ncbi.nlm.nih.gov/books/NBK1154>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28senataxin%5BTIAB%5D%29+OR+%28SETX%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- SENATAXIN  
<http://omim.org/entry/608465>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SETX.html](http://atlasgeneticsoncology.org/Genes/GC_SETX.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SETX%5Bgene%5D>
- HGNC Gene Family: UPF1 like RNA helicases  
<http://www.genenames.org/cgi-bin/genefamilies/set/1169>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=445](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=445)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/23064>
- UniProt  
<http://www.uniprot.org/uniprot/Q7Z333>

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